



# GENE NEWS

*A Publication of the Hawai'i Department of Health Genetics Program*

## HAPPY NEW YEAR FROM THE GENETICS PROGRAM

The Genetics Program staff would like to wish everyone a Happy and Healthy New Year!! This is going to be a busy and exciting year for us. April 2003 is the 50<sup>th</sup> anniversary of Watson and Crick's discovery of the double helix structure of DNA. We are already planning activities to help commemorate this special event. Please watch your mail or check our website ([www.hawaiigenetics.org](http://www.hawaiigenetics.org)) for more information.

### GENETICS FOR YOUR PRACTICE SPRING CONFERENCE 2003

Honolulu, Hawaii

**\*\*\*SAVE THE DATES\*\*\***

**NURSES AND OTHER  
HEALTH CARE PROFESSIONALS:  
PHYSICIANS:**

**Monday, April 14th, 2003**

**Tuesday, April 15th, 2003**



#### TENTATIVE TOPICS:

- **Genetics in Medicine:** Why is Genetics Important for your Practice?
- **Genetics Needs Assessment Results:** What Hawaii's Physicians and Nurses Had to Say About Genetics.
- **Genetic Etiology and Interventions for Common Disorders, including:**
  - Breast, Ovarian and Colon Cancers
  - Cardiovascular Disease
  - Type 2 Diabetes
  - Congenital Hearing Loss
- **Expanded Newborn Screening:** The state adds 15+ disorders to the screening panel
- **Interactive Continuing Genetics Medical Education:** Exploring the new CD-Rom for Genetics Provider Education.
- **What's a Doctor/Nurse to do?** Screening Tools for Genetic Risk Assessment

*For details, check out [www.hawaiigenetics.org](http://www.hawaiigenetics.org), or call Allison at 733-4998.*

*To request a brochure, please call Sharon or Pauline at 733-9055.*

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## Type 2 Diabetes Gene Linked To Heart Disease

A study reveals that there is a close association between a diabetic patient's haptoglobin phenotype (the expression of their gene) and their risk of developing cardiovascular disease. Researchers examined the genetic makeup of individuals from a sample in the Strong Heart Study, a study of heart disease in Native Americans. The study included 206 diabetics and 206 controls between ages 45-74. Although the gene has two forms which are present equally in the general population and diabetics, results revealed a five-fold increased risk of cardiovascular disease in diabetics with one form of the gene over those with the other form. Researchers believe that the study may lead to the development of early preventive measures for cardiovascular diseases in diabetics. *Based on an article published in J Am Coll Cardiol 2002; 40 (11): 1984 – 1990.*

**6.2% of Hawaii's adult population has a form of diabetes.**

*Source: Hawaii State Diabetes Prevention & Control 2001 Data.*



## DNA Testing For Familial Hypercholesterolemia: Improving Disease Recognition And Patient Care

Cardiovascular disease, which is known to have both environmental and genetic components, is the leading cause of death worldwide. Familial Hypercholesterolemia (FH) accounts for 5% of coronary heart disease before age 55. Researchers have found evidence of an association between changes in the gene for low density lipoprotein receptor (LDLR) and the degree of cholesterol lowering by HMG-CoA reductase inhibitors (cholesterol lowering medication). A DNA-based test that helps to identify patients with FH may help in early and cost effective diagnosis, prevention, and treatment. However, further research is needed to better understand the response to these drugs among people with different changes in the LDLR gene. *Based on an article published in Am J Pharmacogenomics 2002;2(4):253-62.*



**FH is inherited in an autosomal dominant manner. It is estimated that 1 in 500 people is affected with FH.**



## Genetics of Congenital Hearing Loss Research Study...An Update

Our Genetics of Hearing Loss Study is anticipated to get underway in early 2003. As we reported in our previous Gene News issue, Hawaii and three other states will be conducting a research study to look at the causes of hearing loss in infants detected through newborn hearing screening or diagnosed with hearing loss before the age of three. The study will involve a full genetics evaluation, genetic testing, and discussion with a genetic counselor and geneticist (a doctor who specializes in genetics).

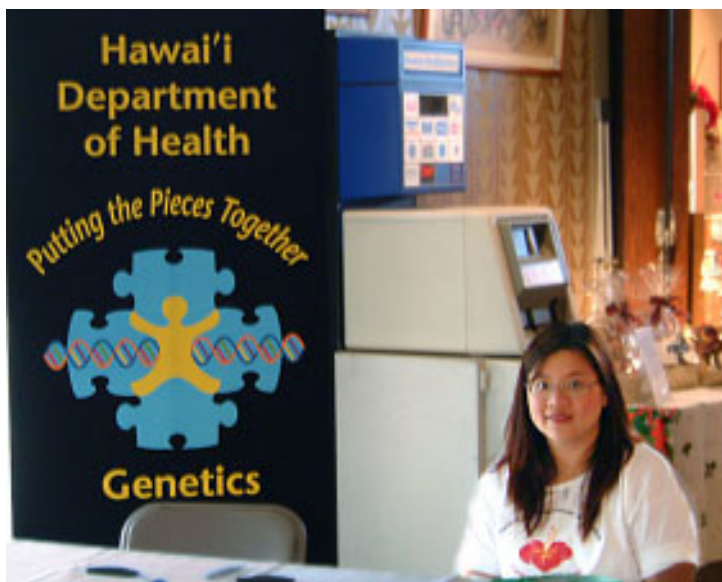
*Please contact the study's genetic counselors for more information:*

Allison Taylor 733-4998

Lianne Hasegawa 733-9039

For more information about the Genetics Program or any of our projects, please go to our website at:

**[www.hawaiigenetics.org](http://www.hawaiigenetics.org)**



We would like to welcome Pauline Mui to our Genetics Program staff. Pauline is a project assistant for the Newborn Screening Tandem Mass Spectrometry Project. In addition to obtaining informed consent at Kapiolani Medical Center for Women and Children, Pauline is busy keeping up with all the administrative details for the project.



## BISHOP MUSEUM

Dogs! Doughnuts! DNA! The Bishop Museum's *Genetics!* Exhibit had it all. The traveling exhibit, on loan from the Pacific Science Center, was at the Bishop Museum from October 5 through January 5. The exhibit is a fun, hands-on collection of displays that encourages people of all ages to discover the exciting world of genetics.

On Sunday, December 8<sup>th</sup>, the Hawaii Genetics Program, in conjunction with the Bishop Museum, Island Rhythm 98.5, and Star 101.9, sponsored a "Genetics Fun Day" to promote the exhibit and to raise awareness of the program and relevance of genetics in everyday life. The fun-filled day included free admission for all children under 12 and families with twins, hands-on genetics displays, games, prizes, bouncy houses, and a lot of educational fun!



*Two future scientists at work in the Genetics Laboratory during the Genetics Fun Day.*

## PROGRAMS IN THE NEWS

The Genetics and Newborn Metabolic Screening Programs have received a lot of positive media coverage lately. In addition to the radio advertisements and community calendar notices for the "Genetics Fun Day" at the Bishop Museum, the Genetics Program was featured in a half page article in the November 21<sup>st</sup>, 2002 Star Bulletin's "To Your Health". The article entitled "*State Aims to Raise DNA Dialogue*" gave a brief overview of some of the program's projects and promoted the "Genetics Fun Day".

The November 25<sup>th</sup> edition of the Star Bulletin featured an article on the Newborn Metabolic Screening Program's identification of a baby with a rare metabolic disorder and an explanation of the expanded screening pilot project at Kapiolani Medical Center. The article was entitled "*Isle Newborn Screening Finds Rare Baby Defect*". The Honolulu Advertiser also ran a similar article "*Newborn Screening Extended*" in the December 15<sup>th</sup> edition and an editorial on the expanded screening in the December 17<sup>th</sup> edition.



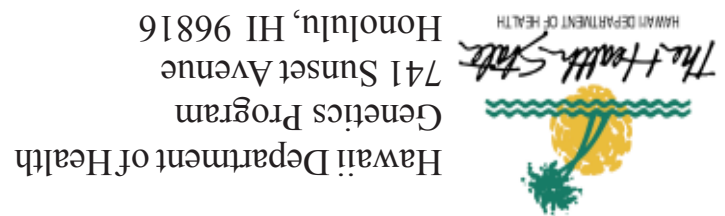
*From Left: Sylvia Au, Lianne Hasegawa, Nicole Sameit & Allison Taylor (of the Genetics Program)*

## Birth Defects Program Wins Recognition

In their 1999 report, "Healthy from the Start", the Pew Environmental Health Commission at the Johns Hopkins School of Public Health gave the Hawaii Birth Defects Program (HBDP) an "A" grade, designating it as one of the top eight programs in the country. The HBDP was one of only four programs nationwide to meet all seven grading criteria. In a 2002 report entitled "Birth Defects Tracking and Prevention: Too Many States Are Not Making the Grade" prepared by the Trust for America's Health, a Washington, DC based advocacy group, the HBDP was recognized as one of eight programs nationally to receive an "A" grade. The HBDP was also cited in the report as being one of only four states to meet all four major standards and 20 sub-criteria for surveillance tracking, data use, prevention and research activities, data sharing, and legislation/adequate resources.

*For more information about the Hawaii Birth Defects Program, please contact Ruth D. Merz at 587-4120 or at [hbdp@crch.hawaii.edu](mailto:hbdp@crch.hawaii.edu)*





## **Tandem Mass Spectrometry Update: Exciting Changes to Our Newborn Screening Program**



We are excited to report that, on October 21, 2002, the Newborn Screening Advisory Committee unanimously voted to support expanding newborn screening using Tandem Mass Spectrometry (MS/MS). The Advisory Committee is comprised of representatives from the statewide birthing facilities, health insurance providers, consumers, public health staff, and health care providers. This expanded screening is expected to begin statewide in September 2003.

Hawaii currently screens newborns for seven different disorders. The expanded newborn screening will use MS/MS to increase the number of disorders to at least 25. The cost of screening will increase slightly to cover the additional screening, diagnosis, and follow-up of the supplemental disorders; traditionally, the cost of newborn screening is covered by insurance.

Expanded newborn screening has been successful in the ongoing MS/MS pilot project at Kapiolani Medical Center. To date, two babies (out of approximately 4,156 screened since March 1, 2002) have been diagnosed with rare metabolic disorders not screened for in our panel of seven. One of the infants was diagnosed with isovaleric acidemia; the other was diagnosed with a variant form of homocystinuria. Both disorders are very rare and are caused by a deficiency of a particular enzyme that breaks down protein. If the disorders had not been detected, these babies could have become very sick and may have died. However, with the help of early diagnosis through expanded newborn screening and appropriate management, both infants are doing well!

*For more information about expanded newborn screening, please contact Chris Matsumoto at 733-9069 or Lianne Hasegawa at 733-9039.*